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Localising genetic testing and screening in Cyprus and Germany

Contingencies, continuities, ordering effects and bio-cultural intimacy

Stefan Beck

Jörg Niewöhner*

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*joerg.niewoehner@hu-berlin.de; ORCID: 0000-0002-9034-9761

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Introduction

Genetic testing and screening comprise diverse fields of practice. They are being employed in medical and public health practice, in different fields of scientific research, in criminal investigations as well as in paternity testing and people's attempts to determine their ancestry. They entangle individuals, families or populations at specific points in time, at specific stages during individual lives, and they follow different ends and produce outcomes, the interpretation and consequences of which are highly contingent upon the specific cultural, social, biological and technological constellations within which they take place (cf. Löwy and Gaudillière 2008). The diversity of these constellations depends to a significant degree on the way they are engaged with and positioned by a multitude of knowledge practices from science and beyond. We present only two examples from the medical domain in this paper and thus ask our readers to take this piece as a point of departure for their own thinking rather than expecting a comprehensive overview of current scholarly and practitioners' debates.

Technology and terminology

Genetic testing is aimed at identifying variants of genes that are associated with inherited disorders. The result aims at confirming or ruling out conditions or at helping to determine a person's risk to develop a genetic disorder or to pass on a trait. In contrast, screening aims to identify individuals in a given population who are at higher risk of having or developing a particular disorder.¹ Thus a genetic test forms the basic biomedical practice for an individualised diagnosis, while screening is one of many specific social settings within which a genetic test is employed. Screening started in the 1960s as a search for 'inborn errors of metabolism' (US PCB 2006). Phenylketonuria (PKU) is seen as the

¹ See NIH 2007 for a detailed version of these definitions: http://ghr.nlm.nih.gov/handbook/testing/genetic_testing (accessed 27 August 2007).

prime example. In general, tests are least controversial when employed in constellations where individuals are able to give their informed consent to the test (NIH/BIG 1998), and where a clearly determined outcome can be expected or the possibility for a positive therapeutic intervention exists (O'Neill 2001).² These preconditions are seldom satisfied *in toto*: individuals may be under the legal age of consent or they may be unable to relate their opinion (Wertz et al. 1994); tests more often than not give probabilistic outcomes and tests support diagnoses of as yet incurable diseases (Evers-Kiebooms 1995). Further, the complexity of genetic knowledge problematises the notion of *informed* consent (Thomson 1994), while the fact that genetic information often pertains to relatives of the person tested questions the notion of *individual* consent (Dillard and Tluczek 2005). Most Western societies have instituted a 'right not to know' in their legal-regulatory apparatus. Decisions concerning one's own genetic constitution are seen as part of an individual's basic right to informational self-determination and are as such protected against undue interference from third persons and the state. Particularly in newborn screenings, this legal constellation makes informational management a highly controversial and complex matter. Lastly, genetic analyses, particularly as part of screenings, often confer *carrier status* onto people, i. e. they confront healthy individuals with the information that they carry a mutation in one of their chromosomes (Clarke 1997; Marteau and Anionwu 1996). This status is unique to *genetic* diagnoses and for many reasons difficult to interpret for those concerned (Ciske et al. 2001).

Case studies

Given the range of issues associated with testing and screening as practices, exemplary case study analyses cannot aim for representativeness in any meaningful sense. Rather, we have selected the thalassaemia screening in Cyprus and the attempts to install a newborn screening for cystic fibrosis in Germany, because they illustrate with particular poignancy the influence of specific local historical and social constellations on the material and discursive practices within which genetic technologies are enacted. We aim at demonstrating that contrary to still influential deterministic conceptualisations that conceive of biomedical technologies as somehow non- or pre-social artefacts that have hegemonic effects (e. g. Winner 1980) on local contexts and ways of implementation, biomedical technologies and practices are co-constructed. They are contingent on past experiences and socio-cultural paths; what screening or testing *is* and what '*effects*' it might have is contingent on its socio-cultural commissioning in specific spatio-temporal contexts.

While the two cases illustrate this point, they could not be more different: in Cyprus, overwhelming support for a population-wide screening results from a highly prevalent disease and a screening regime which has been successfully translated into an existing social structure. In contrast, the deep-seated scepticism towards any kind of 'genetic technology' in Germany arises from the traumatising consequences of state-organised

²For further detail, see the Wilson–Jungner criteria for appraising the validity of a screening programme: www.gp-training.net/training/tutorials/management/audit/screen.htm (accessed 27 November 2007).

eugenic practices during the Third Reich, which continue to shape current modes of public debate, knowledge production and regulation in the domain of medical genetics.

We attempt an anthropological analysis of these two cases and argue against the dominant *exceptionalism* that characterises most critical analyses of genetic testing and screening. Instead, we situate these two cases in the *longue durée* of multiple continuities: of genetic science, of patterns of meaning-making and of regulatory practice. Further, we situate genetic testing practices in the broad continuum of conventional medical testing as an established social practice. In the Cypriot case, we use the notion of bio-cultural intimacy to understand the importance of collective means of coping with genetic diseases and to discuss the effects for collective identities and self-reflection. In the German case, we concern ourselves with the power of collective memory practices to shape the critical analysis of genetic practices. We conclude on a methodological note arguing that the investigation of the interdependencies between bodies and cultures *as practice* necessitates a symmetrical epistemological stance.

Thalassaemia in Cyprus

Cyprus has one of the highest incidences in the world of the mutations that cause β -thalassaemia: every seventh person in the population is a carrier of the trait and suffers from thalassaemia-minor. These heterozygous carriers are generally healthy, but show symptoms of mild anaemia. However, there is a 25 per cent chance that two carriers pass on their respective genes to their offspring. In those cases of homozygosity or compound heterozygosity for a β -thalassaemia mutation, the child will develop thalassaemia.³ In 93 per cent of these cases thalassaemia-major as a very severe and lethal form develops, while only in the remaining 7 per cent of cases patients with thalassaemia-intermediate can lead a life without the need of major therapeutical interventions. According to the carrier frequency in the Cypriot population, almost one in every 160 newborns is expected to suffer from β -thalassaemia-major. The condition usually becomes manifest during the first year of life and – if untreated – leads to a series of severe clinical symptoms. Thalassaemia does not have a specific molecular correlate but includes several clinical abnormalities due to highly ineffective erythropoiesis. Most prominent symptoms are iron overload of the tissue, progressive dysfunction of liver, heart and endocrine glands, enlarged bone marrow resulting in an erosion of the bone structure from within and in pathological fractures. In the skull bones these changes transform the facial features (Olivieri 1999; Weatherall and Clegg 2001).

Starting from the late 1940s, treatments were developed in Great Britain, the US and Australia that reduced suffering and extended life expectancy of patients significantly. Most crucial are regular blood transfusions. However, high transfusion rates that keep haemoglobin levels in normal range contribute to the accumulation of iron overload in patients, that in turn will result in a number of serious health problems. In the late 1960s and 1970s, the optimisation of treatment regimes combining regular blood

³We omit here the symptomatic complexities that arise when different mutations are combined (Weatherall and Clegg 2001).

transfusions with daily intramuscular injections of Desferrioxamine, an iron-chelating agent, successively increased life expectancy to the mid-forties (Modell et al. 2000).

The treatment's high intensity was not only a grave burden for patients and their families but also strained the resources of health care providers. While affluent countries could financially afford to provide the treatment facilities and resources, Cyprus encountered acute difficulties in implementing the new treatment regime: the island had gained independence only in 1960 after a long, violent struggle against British colonial rule. But the new state was troubled by intercommunal conflicts and bloody fights between a minority of Turkish- and a majority of Greek-speaking Cypriots that resulted in a first military intervention of Turkey in her role as guarantor power of the new state in 1964. Subsequently, the separation of communities was proposed as a way of 'solving' the conflict; consequently most Turkish Cypriots were pressed to resettle into 'ethnic enclaves' that were monitored by a United Nations peacekeeping force. In 1974, a coup by right-wing Greek Cypriot militias, intended to unify Cyprus with Greece, provoked an invasion of Turkish troops to protect the Turkish-speaking minority; the following fight by Greek-Cypriots from the northern part of the island and the exodus of Turkish-Cypriots from the South into the Turkish-occupied North completed ethnic cleansings and expulsions resulting in the partition of the island into a 'Turkish' and a 'Greek' part.

These political, social, economical and cultural disruptions provide the context for the difficult implementation of a thalassaemia prevention programme in Cyprus that – in its initial phases – was characterised by lack of funding, hospital facilities, experts, medication, blood supplies and – most important – a general lack of knowledge in the population. As the then leading haematologist, Dr Minas Hadjiminias, recalls:

In the early 1960s there were mothers who suffocated their thalassaemic children with pillows, parents committed suicide and many marriages split after the birth of ill children; because of stigmatisation, families avoided contacts with neighbours, fathers found it difficult to go to the village *kafenion*. We had to fight poverty, ignorance, prejudice, and superstition – not only in patients but in physicians and nurses as well.⁴

The success of the treatment regime despite this difficult situation meant longer treatment, and so it became more and more difficult to find enough blood donors for the growing number of living patients. In addition, the available supplies for the expensive medication with Desferrioxamine were running short in the country, so that families of patients had to buy it abroad (Book 1980: 11). The successful transfer of the advanced treatment regime to Cyprus gradually eroded the capacities of the health care system.

In 1976, two years after the violent political and social events that had gravely disrupted the provision of treatment for patients, Patricia A. Book, a medical anthropologist, conducted fieldwork at the Cypriot Centre for Thalassaemia Treatment and Prevention in Nicosia. The unbearable situation is apparent in her vivid description: On a typical

⁴This passage and other information on the early phases of the thalassaemia programme in Cyprus are based on a series of biographical interviews – conducted by SB – with involved physicians, scientists and representatives of patient groups; here the quote is from Minas Hadjiminias, MD, 2004.

clinic day, the specialised physician, who had received his training in Great Britain, saw 14 patients and was assisted by two practical nurses. Since patients did not have fixed appointment times,

they generally crowded in the hallway at 8.00 a.m., waiting to be called in to see the doctor. Crying, screaming, apprehension, and fear characterized the attitudes of many [of the often very young] patients . . . Some parents reported that they had to bribe, sneak, and/or coax their child or children to the hospital. (Book 1980: 10)

For most of the patients and the accompanying parents, clinic days meant travelling to the hospital over great distances, long waits, distress and fear – twice a month. In addition, because of the shortage of stored blood supplies in Cyprus, parents of thalassaemia patients had to find donors for one to two pints of blood for each child every month of that child's life.

What is obvious from Book's descriptions is that the suffering of thalassaemia patients, of their parents and families, was immense; they had not only to cope with a chronic and fatal disease, but they had to do so in a social environment that was indifferent or even hostile. In addition, families of patients were stigmatised or blamed by co-villagers; thalassaemic children were harassed in school or by their peers because of their facial features or the other symptoms they suffered. Health authorities as well as the leading physicians were alarmed, too. Asked for external advice, the World Health Organisation (WHO) predicted a 300–400 per cent increase in blood requirements and a 600–700 per cent rise in the cost of treatment for the next 50 years, should the birth rates remain unchanged.⁵ In short, the success in the treatment of thalassaemia patients was threatening the very existence of the Cypriot health care system (Angastiniotiset al. 1986: 292). In close cooperation with local experts, the WHO therefore recommended the implementation of a prevention programme to reduce the number of newborns with thalassaemia by means of a premarital carrier screening and a concomitant educational initiative focusing especially on school children.

The reasons for this strategy were medical as well as socio-cultural: while the detection of heterozygotic carriers of the thalassaemia trait is rather simple (measuring red-cell indexes), the detection of homozygotes via prenatal diagnosis was both technically difficult and considered high-risk in the 1970s. Also, in cases of abortion, the procedure put immense psychological as well as physiological stress on women and their families. What was seen as even more problematic was that only after the birth of a first thalassaemic child, when the 'problem' had become apparent, could physicians offer counselling and prenatal diagnosis for further family planning. In contrast, a prospective diagnosis, implemented before young couples had any affected children, was seen to provide a more effective and less stressful point of intervention.

There also existed specific socio-cultural reasons that made early screening and information of carriers a crucial point from the perspective of physicians: cultural modernisation

⁵The treatment costs of a thalassaemia patient from birth to 30 years are calculated to exceed £ 250,000 (Gill and Modell 1998: 761).

and liberalisation of society combined with the economic hardships of post-independence, post-civil war and post-invasion Cyprus had created a new ‘custom’: younger couples after church-authorised engagement were allowed to live together – usually in the home of the bride – until the obligatory dowry could be accumulated and official marriage was possible (Loizos 1975). This praxis of ‘co-residence’ before marriage, however, resulted in severe complications if a premarital screening after engagement eventually showed that both spouses were carriers of the thalassaemia trait. In those cases, there was a high risk that engaged couples broke off their relationship, leaving the bride – being no longer a virgin – with reduced or ruined chances for marrying. Parents of girls therefore tended to resist any recommendation for a screening to prevent the bride and her family from stigmatisation. From the perspective of the medical authorities, a screening before the engagement seemed the only instrument to overcome the opposition to the test.⁶

An educational campaign, targeting school children as well as the general population, also served another purpose that the physicians regarded as eminent: tenacious folk belief held that thalassaemia was likely to be a retribution for past sins of family members. Accordingly, many patients and their families suffered from stigmatisation, often husbands and wives blamed each other or their respective families for having ‘caused’ the disease and in rarer cases children were even isolated in the homes to conceal their existence. These frictions tended to reduce the compliance with the treatment regime so that the leading physicians of the Thalassaemia Centre felt obligated to react (Beck 2007). From their point of view, fighting superstitions and lack of knowledge was the crucial step, and a population-wide screening would have had the added benefit of demonstrating that the thalassaemia trait was widely distributed in the population, also reducing the danger of stigmatisation. The population screening-cum-educational campaign aimed at nothing less than a double reversal: to highlight what was invisible before – namely, that the asymptomatic trait was widely distributed in the population – and to collectivise health problems that were previously considered to be individual or familial.

To achieve their goals, the physicians framed thalassaemia as a collective, ‘Cypriot’ problem and forged an alliance of quite heterogeneous actors: patient groups, politicians, international experts, as well as the leading clergy of the Greek Orthodox Church were commissioned for the prevention programme. While these participants all followed specific interests, the alliance consented to introduce a system of compulsory carrier screening and counselling to prevent carriers from marrying. In addition, a public education campaign was launched using media, schools and different social and cultural organisations; young adults were offered a carrier test. The cooperation with the Church was crucial, because all couples requiring the Church’s blessing for engagement and subsequent marriage have to present a certificate showing that they have been tested and counselled for carrier status. The test results, however, remain confidential, so that two carriers may marry. But since civil marriage has not been a legal option in Cyprus until recently, the testing programme was in fact compulsory for all marrying couples.

The compulsory screening and subsequent information of carriers purposely and effectively added a new criterion to the deliberations in the context of arranging marriages.

⁶Biographical interviews with Michalis Angastiniotis, Minas Hadjiminias; cf. also (Book 1980: 16f).

Until well into the 1980s, marrying in Cyprus involved a complex interplay of rational arrangements, moral and economic evaluations as well as emotional affections between both families and the prospective couple. ‘Love’ appeared rather as a result, not as the cause of a successful marriage: Traditionally, parents arranged marriages within their village to ensure that the combined property of the couple could provide a subsistence basis sufficient for the future family. Arrangements were often made without the knowledge and consent of the young people concerned. Since the early 1970s, however, first young men, then the girls as well, have been able to veto the decision of the parents (Beck 2005; Loïzos 1975). Among the criteria applied for decision-making, the economic, social and moral status of the respective families was the most important; both families would need to very closely scrutinise the other’s economic and social standing, which entailed negotiations to be pursued confidentially and in secret. After all, a marriage candidate turned down could mean loss of face for the entire family (Loïzos 1975: 517). In the agrarian society of Cyprus where conspicuous consumption until recently was largely impossible, the marriage market served as the primary arena for social distinction, a function that was preserved in the following years even under conditions of economic progress and love-marriage (Argyrou 1996).

The pre-engagement screening provided – and still provides – an ‘obligatory passage point’ (Callon 1999) where health professionals have the opportunity to influence reproductive decisions of the young couples. To be sure, two carriers may marry if they decide to do so, and some do,⁷ but usually they make sure that their children are healthy by early prenatal testing and selective abortion. The ‘success’ of this educational campaign and the established public health programme has not only reduced the number of children born with thalassaemia in Cyprus virtually to zero (Angastiniotis and Modell 1998). It even has a remarkable impact on the British health care system, where many young Cypriot migrants who are intending to marry are demanding to be tested for the trait; 98 per cent of all British Cypriot couples in Britain are undergoing premarital testing on a completely voluntary basis (Gill and Modell 1998; Modell et al. 2000). The obligatory screening and counselling for thalassaemia in Cyprus is one of the most successful public health programmes – but it is also arguably the most criticised in the international bioethical debate, mainly because the screening is compulsory and violates the ‘right not to know’ (Hoedemaekers 1998). This accurate bioethical critique, however, does not take into account that the programme was specifically designed to better a public health situation that was perceived as unbearable, to destigmatise thalassaemia patients and their families, to overcome superstition and to provide carriers with a choice in a situation of discrimination. Also, the bioethical critique does not take into account why the programme is still unanimously accepted in the population more than 30 years after its inception.

⁷Couples, where both partners are heterozygotes, tend to have fewer children (up to 20 per cent) than to be expected (Angastiniotis and Modell 1986). See also Petrou et al. (2000).

Newborn screening for *Mukoviszidose* (cystic fibrosis) in Germany

Cystic fibrosis (CF) is a recessively inherited chronic disease that affects the lungs and digestive system. It causes the body to produce unusually thick, sticky mucus that clogs the lungs, leads to lung infections, obstructs the pancreas and stops natural enzymes from helping the body break down and absorb food (Bush 2006). Worldwide, around 70,000 people are affected.⁸ Ever since the ‘cause’ of cystic fibrosis was located in mutations of a gene on chromosome 7 in 1989 (Kerem et al. 1989; Riordan et al. 1989), the genetics of the disease have been a site of great hope, disappointment and controversy (Holtzman 1992). Today, more than 1,300 gene lesions have been deposited in the CF database (Ferec et al. 2006). Among those afflicted that are classified by medical practices as of Caucasian ancestry,⁹ about 70 per cent have a mutation referred to as $\Delta F508$ (Turcios 2005). The picture differs for people classified as of non-Caucasian ancestry.

People who are homozygous with respect to relevant mutations will suffer from the disease, though its progression varies markedly between individuals. Carriers, i. e. people who are heterozygous, will be phenotypically asymptomatic and healthy, but will have a 50 per cent chance of passing on the mutations to their children. Increasing evidence pointing to the benefits of early treatment as well as its cost-effectiveness has led to the introduction of newborn screening (NBS) programmes in many Western countries.¹⁰ In 2007, most countries operate either one nationwide or a large number of regional programmes. The majority of programmes today employ a three-tier test sequence: a biochemical analysis of the level of a particular enzyme (immunoreactive trypsinogen or IRT), a DNA analysis to detect a certain number of mutations and a diagnostic sweat test, measuring the amount of salt in the patient’s sweat (Stern 1997). While the first tier is always an IRT measurement, some variation exists thereafter regarding the type of test and its sensitivity. A survey of 26 programmes in Europe shows that 19 employ mutational analysis, with a median of 31 mutations covering a median of 82 per cent of mutations in the screened populations (Southern et al. 2007).¹¹ Carrier identification and related issues of informed consent as well as the rate of false positives have been intensely debated over the last 15 years (Decruyenaere et al. 1998; Fries et al. 2005; Parsons and Bradley 2003; Watson et al. 1991). Treatment of the condition involves predominantly dietary changes, physio- and breathing therapy, as well as the use of mucus-dissolving drugs combined with autogenic drainage. Improvements in treatment have meant that sufferers now have an average life expectancy of 37 and rising (Davis 2006).

In Germany, where the disease is commonly referred to as *Mukoviszidose* from the Latin *mucus* for phlegm and *viscidus* for viscous or clingy, a national newborn screening

⁸Current figure from the Cystic Fibrosis Foundation. See www.cff.org (last accessed 22 November 2007).

⁹We recognise that the classification ‘Caucasian’ is problematic in many ways (M’charek 2005). Throughout this text, this terminology does not reflect our own perspective but the dominant narrative in evolutionary biology and medicine.

¹⁰As screening programmes have not been running for long enough to show an effect on life expectancy, benefits have been discussed controversially (Koscik et al. 2005; Rock 2007).

¹¹NB: It is Austria, Northern Ireland and some of the Italian regional programmes that operate without a genetic component (Southern et al. 2007).

covered by the national health service has so far not been introduced.¹² A number of clinics throughout the country offer the service to those willing and able to pay for it. In 2008, the administrative body with the power to grant national health service approval seems to be close to commissioning a formal cost-benefit analysis. Provided this confirms the international status quo, approval may be granted and introduction into the standard state health sector programme may proceed. This is likely to take considerably more time and debate.

The main patient organisation, the Mukoviszidose e.V. – the federal representative of several regional groups referred to as *Regios* – has been playing a very active role over the last 40 years. Apart from providing a point of contact for those concerned, the group funds applied biomedical research currently with € 2.5m for a programme running 2008–10. In 2006 the organisation also founded the Mukoviscidosis Institute – a non-profit, limited company – in order to initiate, provide support for and coordinate research, particularly (pre-) clinical trials and epidemiological surveys. The organisation is staffed *inter alia* with clinicians, and over the last few years has been lobbying for the introduction of a nationwide newborn screening on the basis that benefits of early treatment outweigh the risks, particularly to carriers. Large active patient groups such as described for other European countries or North America (cf. Callon and Rabeharisoa 2008) are still relatively unusual in Germany. The Mukoviszidose e.V. together with the German cancer relief programme Deutsche Krebshilfe e.V. form notable exceptions.

In order to try to understand why Germany continues to be so reluctant to introduce a newborn screening programme despite an emergent international consensus, it is helpful to look in more detail at an existing screening programme at the obstetric clinic of the Technical University Dresden (TUD) in the federal state of Saxony – the only programme we know of which has secured outside funding to offer a screening to all couples free of charge at the point of service.¹³ The clinic has offered this screening since 1996 and continues to do so, but it has never lost the status of a pilot project. From 1976 to 1985, the TUD, then an institution of the German Democratic Republic, screened for cystic fibrosis as part of a newborn screening for phenylketonuria (PKU). This programme was terminated in 1985. In the early 1990s, after German reunification, the number of children with *Mukoviszidose* who presented at the obstetric clinic in Dresden at the very late age of five and six in very serious condition increased perceptively. The local clinicians, who had been involved in the early screening programme, were not prepared to put up with what they perceived as an unacceptable decline in standards of care and began to rally for support. By June 1996 they had succeeded in raising enough money from a number of predominantly public sources to begin a new screening programme. Following international standards, the screen is offered to all parents conditional upon their informed consent, which is obtained prior to birth. Full results including carrier status are communicated to the parents, extensive counselling is offered and the integration into the specialised cystic fibrosis care centre is arranged for affected children.

¹²Information gleaned from interviews conducted by JN throughout 2007.

¹³See www.neoscreening.de/DGNS/frame_Website.htm for further information (accessed 11 November 2007).

On the surface, this is a trivial story. It speaks of a bureaucratic administration that is possibly a little slow in following an emerging international consensus on scientific evidence, cost-benefit and best practice. A policy network analysis might reveal the often-divergent interests of lobbying groups and the complex structure of the German self-governing health system. Beneath the surface, however, it is the finer details that reveal the highly German specificity of this case that are instructive for the analysis of some rather less trivial issues.

The Dresden programme informs parents of their child's carrier status. Approval by the relevant ethics commission for this procedure's informed consent protocol was granted in the 1990s. It is clear from our work that, today, several attempts in other federal German states to integrate programmes into standard care have serious difficulties in getting ethical approval for their respective protocols. This is mainly due to the German Society of Human Geneticists' guidelines emphasising the right not to know, particularly for carriers:¹⁴ children ought to be protected from this information until they reach the legal age of consent. Two kinds of consequences arise from this stipulation: (1) the clear-cut distinction between DNA-based tests and conventional diagnostics speaks to fundamentally different frames of reference, namely human dignity in the case of DNA tests and pragmatics in conventional diagnostics; (2) procedurally, many attempts are being made at circumnavigating the bioethical frame, e.g. via lowering IRT thresholds in order to be able to invite child and parents to a sweat test without having to refer to a positive DNA result. The extreme sensitivity towards the ethical frame of reference means that informed consent procedures usually initiated in the third trimester of pregnancy have become central to screening discussions. In order to satisfy ethical review boards, informed consent forms often run into five to six pages of fine print. This increases parental insecurity and often leads to a refusal of the test without even reading to the end of the document. From a clinical perspective, the development and the application of such immensely complex and hard-to-use forms is simply judged impractical. As a result, many local screening programmes struggle to get off the ground. Moreover, the TUD programme currently considers substituting the genetic test for a second biochemical analysis (pancreatitis-associated protein assays), which is not 'genetic' and does not reveal carrier status, in order to avoid further difficulties in this area.

This particularly strict reading of informed consent requirements raises issues, in Germany, of clinical pragmatics on the one hand and ethical reflection on the other. Both discourses centre on individual civic rights and moral exigencies. The biomedical-ethical debate aims to develop a protocol that is deemed to protect a universal individual from undue infringements of its basic rights as a human being – for all the right reasons we emphasise. However, this particular focus on an individual ethics sidelines at least one other important issue: screening programmes in major cities such as Berlin, that are inhabited by a significant number of people of non-European origin, need to deal with a large amount of heterogeneity with respect to the number and kinds of cystic fibrosis relevant mutations. A recent survey of European CF centres reports, for example,

¹⁴See Stuhmann et al. (2006) and www.gfhev.de/de/leitlinien/gfh.htm?Submit2=Liste+anzeigen for further information on earlier guidelines.

31 different mutations for Turkish migrants (Lakeman et al. 2008). The mean detection rate of the three most commonly used panels lies at 44.9 per cent and can be expanded to 57.9 per cent when including 13 of these 31 mutations. The sensitivity of expanded tests is judged too low to warrant any kind of screening of Turkish immigrants in European societies (Lakeman et al. 2008: 32). These figures differ from the numbers for patients living in Turkey raising important questions about the reasons for this effect (Schoorl et al. 2001). In the context of this paper, it is of particular relevance that these figures raise questions of human biological diversity and ethnic belonging, the medical construction of a (sub-)population, population-based protocols as well as issues of access to health care and research priorities in the field of migrant health. These are all questions with, *inter alia*, an ethical dimension. Yet in Germany, and possibly elsewhere, these questions are debated in the specialist circles of paediatricians and human geneticists only. Necessarily, this debate is focused primarily on the technical and practical issues of test sensitivity and specificity as well as matters of the various protocols' efficiency and effectiveness, while the political-cum-ethical implications are muted. The *desideratum* of a wider ethical, political and public debate is entirely muted despite the fact that in the large cities, such as Berlin, about a third of the population is considered of non-German origin (just under 4 per cent of Turkish origin).

Discussion

Continuities and ruptures

Genetic tests are embedded in and contingent on material-discursive practices with their own specific historicity. On the basis of our case material, we focus on three specific areas of continuity, connection and rupture. We are aware that this discussion needs to be read with an appreciation of the *continuity of scientific knowledges*, particularly the continuous history of genetics (Müller-Wille and Rheinberger 2007; Rheinberger and Gaudillière 2004), from structuralist and functionalist concepts of the gene to postgenomic notions of 'gening' (Fox Keller 2006; Griesemer 2002; Jablonka and Lamb 2006), which situates the lingering notions of determinism as well as the expectations invested in the technology by medico-technological practices as well as public discourse. Also, genetic tests are almost always embedded in a testing regime for biological information, which spans the continuum from genotype to phenotype. Within such regimes, the usefulness of genetic information is certainly not increasing with expanding post-genomic knowledge about the complexity of aetiologies.¹⁵

¹⁵See also the debate on microarray technologies (Shuster 2007). The nevertheless increasing use of this information is driven primarily by the involvement of the pharmaceutical industry, e.g. via the marketing of drugs targeted to specific mutation profiles (Hedgecoe 2004; Kollek et al. 2004; Meyer 2004).

Continuity of patterns of meaning making and social poetics

Understanding the specific formation of the genetic testing regime for thalassaemia in Cyprus becomes possible only through recognising how the condition had been handled before genetic knowledge as a set of post-war biomedical knowledge practices arrived on the scene. In Cyprus, thalassaemia has always been a highly visible issue. It has been firmly integrated into social and cultural patterns of meaning-making: explanatory models have been attuned to highly localised practices of arranging marriages, organising and understanding kinship and sanctioning breaches of established social norms. It would be misguided to assume that genetic testing has simply been superimposed on this existing constellation. Cypriot medical researchers and clinicians were acutely aware of the possible social and political consequences their knowledge might have. They carefully aimed at integrating these new technological possibilities into existing social practices; and these new options were adopted by non-experts according to their specific rationales. Hence, what unfolded in Cyprus is not adequately described as a straightforward roll-out of a new technology but rather a slow process of translation (Callon 1999) and situated learning (Chaiklin and Lave 1993): not centrally controlled but carefully and ingeniously appropriated by a multiplicity of networked actors. The result is a changed pattern of practice, which now includes premarital screening, pre-implantation diagnostics and prenatal genetic tests (cf. also Franklin and Roberts 2003). Today, after civil marriage without a screening certificate has become legal, the programme is still adopted unanimously: an in effect compulsory, directive and invasive screening programme is legitimated bottom-up by everyday practice. Instead of conceptualising genetic testings and screenings as having preconfigured 'politics' and deterministic 'effects' independent of social contexts, we advocate a perspective that emphasises processes of social poetics (Herzfeld 1997: 139–55) where actors pragmatically integrate new – non-neutral – options with pre-existent cosmologies, practices and institutions.

Political-regulatory continuity

The German case study shows another kind of historical continuity. Here, the collective memory of the Holocaust and the atrocities committed against the disabled by the medical establishment during Nazism have greatly sensitised an entire generation of physicians as well as public discourse and political decision-making (Müller-Hill 2000; Paul 1995) to practices that many commentators today problematise as neo-eugenic (Duster 1990). Thus, for all the right reasons, the involvement of a genetic test as part of a screening programme has immediately triggered an ethical debate. At its centre stands the well-rehearsed discursive figure of the 'slippery slope', i.e. the perceived inevitable dynamic from neonatal to prenatal and preimplantation diagnostic procedures and the subsequent questions of reproductive decision-making and eugenic practices (Ethikrat 2003). The debate is institutionalised at many different levels, and its intensity provoked contributions not only from specialists in the medical or ethical field but also from public intellectuals like Jürgen Habermas who diagnose the advent of a 'liberal eugenics' (Bundestag 2001; Habermas 2001).

This worry about a resurgence of eugenic practices is by no means confined to Germany (cf. Nelkin and Lindee 1997). Yet it is important to note the unique discursive constellation here. Bioethical ‘principlism’ (for a critique, see Jonsen and Toulmin 1988) combines readily with biomedical ‘factualism’, both being impregnated by universalistic reasoning (Honnefelder et al. 2003). More often than not, the ‘factual basis’ of the debates, informed by biomedical perspectives, remains unchallenged (Light and McGee 1998; Turner 2003; Wertz 1998). Questioning this basis and its dynamic, Margaret Lock rightly speaks of the *tenacity of hyperbole* (Lock forthcoming). Hyperbole refers to the visionary rhetoric of an ‘enriched future’ relentlessly emanating from many quarters of biomedical and genetic science – despite the growing realisation of biological complexity emerging from everyday work in the laboratories that precludes ‘simple’ models of genetic determinism. Many, including Lock (Lock 2005), contextualise this tenacity of hyperbole: in the specificity of the scientific field (Bourdieu 1975), in the sciences’ questionable self-conception as modern (Latour 1993) and the economic and institutional dynamics of emerging fields of research (Hedgecoe 2003). In Germany, however, the link into collective memory practices and a historical continuity of regulatory decision-making means that much of the critique and worry immediately attaches to hyperbole – in a reflex-like, powerful link that is hard to question in a public arena. While this is not problematic *per se*, we argue that the bioethical debate in Germany runs the risk of ‘bioethical reductionism’ and thus hinders a thicker understanding of *situated* genetic screenings and testings.

Local ruptures

A brief comparison with the introduction of thalassaemia screening in the UK reminds us that continuity must not be mistaken for smooth progress. Rather, local ruptures can also result: prevalence of thalassaemia amongst migrants of Mediterranean and Pakistani background in the UK was deemed high enough to warrant screening in 1977 (Modell et al. 2000). An information campaign was required that would target this subpopulation. British medical practitioners were very much aware of international bioethical debates as well as the politically sensitive nature of dealing with ethnic minority groups. Hence, voluntary participation, the protection of individual and group autonomy and informational rights as well as informed consent and discrimination concerns played an important role in the set-up of the screening programme. The result was an information campaign targeting all pregnant women of ‘not North European origin’ (Modell 1986: 388) in the country. While this campaign was conceived not to discriminate *unnecessarily against people*, it did not discriminate *enough between populations* for the campaign to have an effect on the targeted people. It was considered a failure according to its inventors because it did not single out and speak to the people who were intended as the audience. While this comparative anecdote by no means warrants ignorance towards bioethical considerations and political sensitivities, it does open an important line of argument suggesting that bioethical positions are not universal in nature but do need to be attuned to the specific local and historical context within which they operate.

Ordering processes

Against the backdrop of these specific continuities and ruptures, which have shaped the translation of particular knowledge practices into local contexts, the following sections discuss the role of genetic testing and screening regimes in altering ordering processes. By *ordering* we mean the manifold classification work (Bowker and Star 1999) which emanates from communities of practice (Lave and Wenger 1991).¹⁶ Many effects of testing and screening have been discussed at the level of the individual – self-consciousness, personhood, corporeality – and the level of discourse regarding concepts of health, illness and normality. In keeping in line with our social anthropological analysis, we focus on the level of sociality, i.e. on the way knowledge practices change collectivising agency and help to shape a sense of belonging, which we discuss below as bio-cultural intimacy and biosociality.

Discrimination and de/stigmatisation

The Cypriot case illustrates how explanations for health and illness are produced in highly localised patterns of meaning-making. Often, these explanations have been invested with strong moral connotations. Suffering from thalassaemia was readily identified as a divine punishment for a breach of established social norms. Afflicted people and their families were stigmatised for their moral failure and shunned by the community. Introducing the screening thus ‘rationalised’ and ‘objectified’ thalassaemia. It set off a process of *Entzauberung*, i.e. the demystification of a phenomenon, and its translation into Western modernist thinking (Weber 1922/1988). The cause of hereditary diseases, then, is considered no longer a *moral* but a *molecular* failure. This reallocation of blame has the potential to destigmatise the individual. Furthering this process is an evolutionary narrative that depicts the ‘problem’ as *collective* fate.

Citizenship

The *tenacity of hyperbole* (Lock forthcoming) precludes a public debate about testing as a situated technology in a specific institutional and cultural setting. What is at stake, however, are the ontological and epistemological dimensions of ordering processes as well as their biopolitical effects. Genetic testing understood within hyperbole confronts those positively tested not with an illness but with a *genetic* disease. In public discourse, this label readily preconfigures the *conditions of possibility* (Foucault 1972) and confers identity. The Mukoviszidose e.V. is trying to escape from this trap by understanding test results not as prefigured genetic knowledge but as genetic information, the interpretation of which is neither certain nor merely a matter of genetic science. Yet they stay within the dialectic of hyperbole insofar as they position themselves relative to many hypothetical ‘if ... then’ scenarios rather than voicing their justified concerns about current medical practice and care.

¹⁶NB: this classification work orders not only what is visible and how (epistemology) but it also stabilises phenomena (ontology) (Barad 2007).

Understanding a genetic test as a part of a *biomedical platform* (Keating and Cambrosio 2000) offers a different analytical angle. It positions tests as a social practice entangled with biomedical technologies and knowledges, patient groups, economies and embedded within a certain cosmology (Herzfeld 1987). A platform thus marks an ordering practice that translates existing meaning-making practices to arrive at new ‘systems of claims and ethical projects that arise out of the conjugation of techniques used to govern populations and manage individual bodies’ (Nguyen 2005: 126) – it thus characterises a biopolitical process, which provokes what has been termed *genetic* or *therapeutic citizenship* (Heath et al. 2004; Nguyen 2005).

German screening debates, on the other hand, are still caught up within hyperbole. Paradoxically, this constellation rests on the very notion of essentialising biological citizenship it is trying to denounce. This dilemma crystallises in the fact that the debate about genetic testing and screening in the Turkish migrant population is framed as an economic rather than ethical issue. Genetic tests here are integrated into a biomedical platform, which makes it difficult to handle biological-cum-ethnic-cum-cultural difference. The North American model of culturally sensitive care has an increasing tendency to produce ethnicity as a readily accessible marker for biological difference, thus biologising and geneticising cultural difference (Duster 1990; Duster 2006; Lipphardt and Niewöhner 2007; Niewöhner 2007). The German health system, on the other hand, produces problematic injustices through ignoring biological difference altogether.

Bio-cultural intimacy, biosociality and the gene pool as a ‘tragic commons’

This last aspect takes the analysis a little beyond clinical practice and into the changing scientific discourse on thalassaemia and cystic fibrosis. In both cases, the respective genetic mutations are thought to provide resistance against malaria and a number of gut infections, respectively. It is hypothesised that the differences in prevalence in diverse populations are the outcome of selection processes due to specific environmental conditions – namely malaria and cholera. There are many problems with these kinds of evolutionary narratives, which we cannot discuss here. Irrespective of these problems, however, these widely publicised discourses do contribute to a reworking of *narratives of collective pasts*: they invoke a shared history of migration, adverse living conditions and hardships. Alongside other (f)actors, they create and preserve a sense of what we call *bio-cultural intimacy*. With the notion of ‘cultural intimacy’, social anthropologist Michael Herzfeld refers to those aspects of cultural identity that are considered a source of embarrassment in situations of contact with outsiders. Nevertheless, it provides ‘insiders’ with an assurance of common sociality and serves as a central source of defiant pride, critical self-interpretation or -rationalisation (Herzfeld 1997: 3). Similarly, but stressing the effect of scientific knowledge and classificatory practices, social anthropologist Paul Rabinow (1992; 2007) coined the term ‘biosociality’ to refer to the potentiality of genetic diagnoses to create new identities (e.g. carrier of mutation X), collectivities (e.g. descendants of the first carriers of mutation X) and collective forms of action (e.g. the creation of patient/lobby groups that have a ‘mutation’ as common denominator) (cf. Gibbon and Novas 2007).

While Rabinow's biosociality points to explicit *knowledge, truth* and *action*, Herzfeld's concept refers more to implicit *cultural cosmologies, experience* and *reflection*. We suggest combining both aspects in the term *bio-cultural intimacy* in order to analyse historical, cultural and social contingencies in the way biomedical options are appropriated and embedded into everyday life via meaning-making practices. Applying this perspective, the tension between scientific and everyday knowledges, the potential conflict between scientifically validated truth and experience-based shared convictions, or the clash between hegemonic rationalities and heterodox reasoning can be analysed. Bio-cultural intimacy enables Cypriots to interpret thalassaemia as a collective 'ethnic' fate, to understand the gene pool as a 'tragic commons' that requires collective management that the traditionally weak state is only insufficiently able to provide, and which accordingly requires collective forms of self-knowledge, self-representation and self-intervention. In contrast, the question of how to manage the risk of cystic fibrosis for Germans is understood against the backdrop of Nazi eugenics, discussions about human dignity of 'unborn life' and an ethics of individualism that is implemented in the context of a welfare-state system that (still) grants to all citizens equal access to medical care and diagnosis.

Particularly the latter section of the analysis illustrates the need for concepts that take seriously the dialectic of and the interdependence between biology and culture. *Local biology* (Melby et al. 2005) is such a concept, as it argues that lived bodies are always shaped by the irresolvable interdependence of bios, social practice and local cosmologies (Lock and Kaufert 2001). In the same way that the science behind genetic tests fosters *memory politics* by reworking narratives of a collective past, it also shapes the future by *making up local biologies* (Hacking 1986, 1995) through intervening in human reproduction.

The immediate consequences of this perspective are methodological and epistemological: if thalassaemia is not simply a molecular disease investigated by biomedical methods and if genetic screenings are not simply a social practice investigated by social scientists, then we need to pay attention to the way thalassaemia in Cyprus is produced as a result of a complex material-discursive assemblage (Rabinow 2003) involving ill people, postcolonial sentiments and predispositions, screening technology, labs, traditional marriage practices in transformation, the Church, evolutionary biology, genes, feelings of communality and collectivity, and so forth. Taking this complexity of interacting factors, facts and artefacts seriously means employing methods that are able to symmetrically register material, social and semiotic practices. And we need an epistemology, which sets a different *agential cut*, i.e. a perspective which does not reproduce existing dichotomies of nature and culture but allows materiality and discourse to make their contribution to stabilising particular lived bodies (Barad 2007; Lock 2005).

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